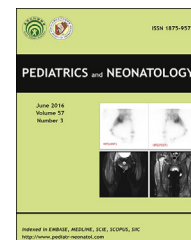


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## EDITORIAL

# Nationwide Population-Based Epidemiologic Study of Hirschsprung's Disease in Taiwan



In 1886, Harald Hirschsprung first described two cases of infantile constipation with colonic dilation, but he did not suggest any etiology or therapeutic option.<sup>1</sup> Over the past 30 years, many studies have revealed the clinical presentation, radiological and histological diagnoses, surgical procedures, epidemiology, and associated genetics related to colonic dilation.

Hirschsprung's disease (HD) is a congenital disease with a defect in the enteric nervous system, leading to significant morbidity and even mortality in early childhood. It is usually subdivided into short- and long-segment disease. In rare cases, there is total colonic aganglionosis and even total intestinal HD. The aganglionic intestinal segments cause mechanical obstruction because of their failure to

relax during peristalsis, which results in intestinal obstruction in neonates and severe constipation in infants and children. Surgical resection of the aganglionic segment and pull-through operation are essential parts of HD treatment.

HD occurs as an isolated trait in 70% of patients. Syndromic HD occurs in 30% of cases, and 40% of them are associated with a chromosomal abnormality, with trisomy 21 being the most frequent. In recent years, significant progress has been made to understand the molecular genetics of this disorder. Several susceptibility genes have been identified, all of which are involved in the development of the enteric nervous system. The known susceptibility genes responsible for HD include *RET*, *EDNRB*, and *SOX10*.<sup>2</sup>

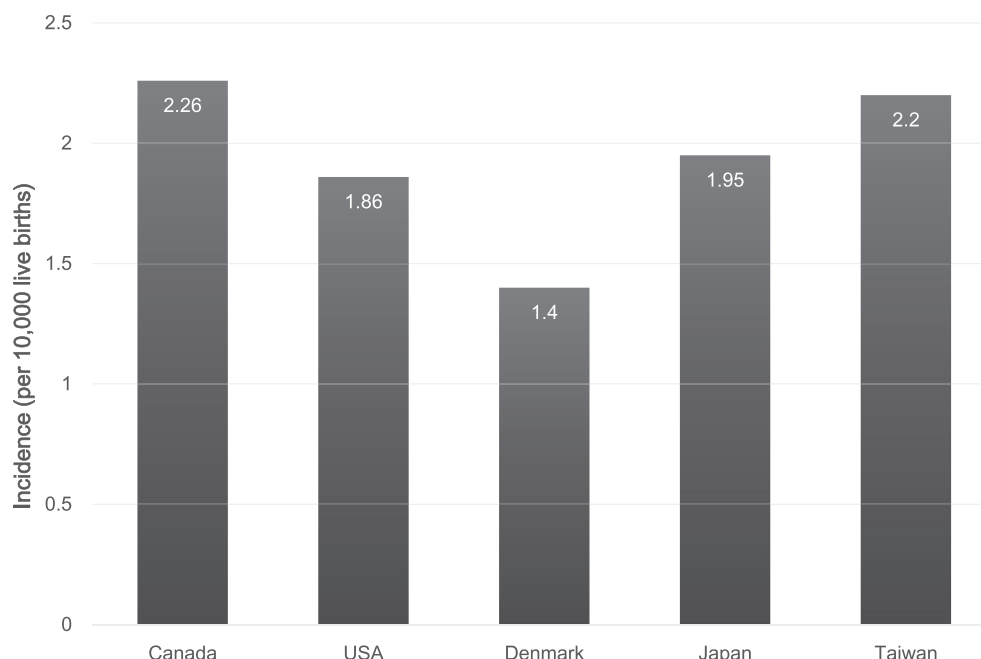


Figure 1 Incidence of Hirschsprung's disease across countries.

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The incidence of HD varies from 1/5000 to 1/10,000 live births (Figure 1).<sup>3–6</sup> There appears to be a male preponderance with a ratio of 3:1 to 5:1, especially in the short-segment disease (80% of HD cases). However, most of the epidemiologic studies have been conducted in the white population, and thus, it is not clear whether there are any interracial differences.

There is a lack of data on the incidence of HD in the Chinese population. In this issue of *Pediatrics and Neonatology*, Chia et al<sup>7</sup> retrieved data for 629 HD patients who received a surgical intervention between 1998 and 2010 from the Taiwan National Health Insurance Research Database (NHIRD). They found that the incidence of HD in Taiwan was 2.2/10,000 live births (male-to-female ratio: 2.38). Among the HD cases, 26.9% had associated anomalies, 24.3% had preoperative enterocolitis, and 26.9% had postoperative enterocolitis. The average medical expenditure for each patient was US \$4513.

However, this study had a few limitations. First, NHIRD contains little information on data that are not necessary for reimbursement claims. The study did not provide pathological information and the extent of aganglionosis, which are crucial to accurately diagnose HD and assess its severity. Second, preoperative and postoperative enterocolitis cases were not reported in enough detail because the diagnoses were made only according to the International Classification of Diseases, 9<sup>th</sup> Revision, Clinical Modification codes in the NHIRD rather than based on patient's medical records. The diagnostic code in the NHIRD is meant for billing purposes and thus may not be accurate. Third, there are no data regarding the surgical procedure performed and the long-term outcomes.

Despite the aforementioned limitations, because the NHIRD contains information on a nationwide population scale, the study by Chia et al<sup>7</sup> is unique for pediatricians and public health researchers to understand the demographic data of HD in Taiwan.

## Conflicts of interest

The author declares no conflicts of interest.

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